DISORDERS SCREENED BY THE PROGRAM

These are disorders which may have significant mortality and morbidity when not diagnosed pre-symptomatically and may not be consistently identified clinically in the neonatal period. Early detection and treatment may improve the health and development of newborns identified with these disorders. More information on these conditions are available on the South Dakota Newborn Screening Website: http://www.state.sd.us/doh/NewbornScreening/

ENDOCRINE DISORDERS:

- Congenital adrenal hyperplasia (CAH) *
- Congenital hypothyroidism (CH) *

CYSTIC FIBROSIS *

HEMOGLOBINOPATHIES*:

- Sickle cell disease and other hemoglobin disorders

METABOLIC DISORDERS:

- Biotinidase deficiency *
- Galactosemia *

Amino Acid Disorders - Disorders identified through tandem mass spectrometry testing, listed with abbreviations and names:

- (ASA) Argininosuccinate acidemia*
- (CIT 1) Citrullinemia or ASA Synthetase Deficiency*
- (HCY) Homocystinuria (cystathionine beta synthetase)
- (MSUD) Maple Syrup Urine Disease*
- (PKU) Phenylketonuria*
- (TYR-1) Tyrosinemia Type 1*
- (ARG) Arginiemia**
- (BIOPT-BS) Defects of biopterin cofactor biosynthesis**
- (CIT-II) Citrullinemia type II**
- (BIOPT-RG) Defects of biopterin cofactor regeneration**
- (H-PHE) Benign hyperphenylalaninemia**
- (MET) Hypermethioninemia**
- (TYR II) Tyrosinemia type II**
- (TRY III) Tyrosinemia type III**

Fatty Acid Oxidation Disorders - Disorders identified through tandem mass spectrometry testing, listed with abbreviations and names:

- (CUD) Carnitine uptake defect (Carnitine transport defect)
- (LCHAD) Long-chain L-3 hydroxyacyl-CoA dehydrogenase*
- (MCAD) Medium chain acyl-CoA dehydrogenase*
- (TRP) Trifunctional protein deficiency*
- (VLCAD) Very long-chain acyl-CoA dehydrogenase*
- (CACT) Carnitine acylcarnitine translocase**
- (CPT-Ia) Carnitine palmitoyltransferase I**
- (CPT-II) Carnitine palmitoyltransferase II**
- (GA-II) Glutaric acidemia Type II**
- (MCKAT) Medium-chain ketoacyl-CoA thiolase**
- (M/SCHAD) Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase**
- (SCAD) Short-chain acyl-CoA dehydrogenase**

Organic Acid Disorders - Disorders identified through tandem mass spectrometry testing, listed with abbreviations and names:

- (GA-1) Glutaric acidemia type 1*
- (HMG) 3-Hydroxy 3-methylglutaric aciduria *
- (IVA) Isovaleric acidemia*
- (3-MCC) 3-Methylcrotonyl-CoA carboxylase*
- (Cbl-A,B) Methylmalonic acidemia (vitamin B12 disorders)*
- (BKT) Beta Ketothiolase*
- (MUT) Methylmalonic Acidemia (methylmalonyl-CoA mutase)*
- (PROP) Propionic acidemia*
- (MCD) Multiple carboxylase*
- (2M3HBA) 2-Methyl-3-hydroxybutyric aciduria**
- (2MGB) 2-Methylbutyrl-CoA dehydrogenase**
- (3MGA) 3-Methylglutaconic aciduria**
- (Cbl-C, D) Methylmalonic acidemia**
- (IBG) Isobutyrl-CoA dehydrogenase**
- (MAL) Malonic acidemia**

* American College of Medical Genetists Recommended Disorders - Core Panel

** American College of Medical Genetists Recommended Disorders - Secondary Targets

Caveat: The possibility of a false negative or a false positive result must always be considered when screening newborns for metabolic disorders.

Table I
Summarizes the disorders screened in South Dakota, including the incidence, symptoms, and treatment.